## CYSTIC FIBROSIS (CF)

### What is it?

Cystic fibrosis (CF) is an inherited disease of the mucus glands that affects many body systems. In particular, this disorder causes progressive damage to the respiratory (lung) system and chronic digestive system problems.

Mucus is a slippery substance that lubricates and protects the linings of the airways, digestive system, reproductive system, and other organs and tissues. In people with cystic fibrosis, the body produces mucus that is abnormally thick and sticky. This abnormal mucus can obstruct the airways, leading to severe problems with breathing and bacterial infections in the lungs. These infections cause chronic coughing, wheezing, and inflammation. Over time, mucus buildup and infections result in permanent lung damage, including the formation of scar tissue (fibrosis) and cysts in the lungs.

Most people with cystic fibrosis also have digestive problems because thick, sticky mucus interferes with the function of the pancreas. The pancreas is an organ that produces insulin (a hormone that helps control blood sugar levels) and enzymes that help digest food. Mucus blocks the ducts of the pancreas, preventing these enzymes from reaching the intestines to aid digestion. Problems with digestion can lead to diarrhea, malnutrition, poor growth, and weight loss. Some babies with cystic fibrosis have meconium ileus, a blockage of the intestine that occurs shortly after birth.

### What are the symptoms?

Infants with CF appear normal at birth and can have a variety of symptoms, including: very salty-tasting skin; persistent coughing, at times with phlegm; frequent lung infections; wheezing or shortness of breath; poor growth/weight gain in spite of a good appetite; and frequent greasy, bulky stools or difficulty in bowel movements. An early diagnosis of CF and a comprehensive treatment plan can improve both survival and quality of life.

### How do you get it?

CF is inherited in an autosomal recessive pattern. As an autosomal recessive disorder, the parents of a child with CF are unaffected, healthy carriers of the condition and have one normal gene and one abnormal gene. With each pregnancy, carrier parents have a 25 percent chance of having a child with two copies of the abnormal gene, resulting in CF. Carrier parents have a 50 percent chance of having a child who is an unaffected carrier and a 25 percent chance of having an unaffected, non-carrier child. These risks would hold true for each pregnancy. All siblings of infants confirmed to have CF also should be tested; genetic counseling services should be offered to the family.

**How is it detected?**

CF may be detected through newborn screening. Confirmation of newborn screening results is required to make a firm diagnosis. The confirmation test done to confirm a diagnosis of CF is a "sweat test". A sweat test measures the concentration of salt in the person's sweat. A high level of salt indicates CF. The test is painless and is done at a Cystic Fibrosis Foundation accredited care center. Some babies will have inconclusive results and need to have additional testing.

**How common is it?**

The incidence of CF is 1 in 2,500 to 3,000 births in Missouri.

**How is it treated?**

Treatments will vary depending on the baby, but will typically include special medicine to help the baby's digestion, a high-calorie protein diet and special chest therapy. The best medical care for babies with CF is at care centers that are accredited by the National Cystic Fibrosis Foundation.

**DISCLAIMER: This information is not intended to replace the advice of either a pulmonologist, who specializes in the treatment of children with CF, or a genetic medical professional.**

**Where can I get services?**

Provision of names listed below does not necessarily include all hospitals or private practice physicians who may treat children with CF. The Cystic Fibrosis Foundation accredited care centers in Missouri are:

Cardinal Glennon Children's Medical Center
St. Louis, MO
314-268-6439

Children's Mercy Hospital
Kansas City, MO
816-983-6628

St. Louis Children's Hospital
St. Louis, MO
314-454-2353

University Hospitals and Clinics
Columbia, MO
573-884-8579

**For more information:**

**American College of Medical Genetics - Newborn Screening ACT sheets and Confirmatory Algorithms**
http://www.acmg.net/resources/policies/ACT/condition-analyte-links.htm

**Cystic Fibrosis Foundation**
http://www.cff.org/

**Genetics Home Reference**
http://ghr.nlm.nih.gov/condition=cysticfibrosis

**Medline Plus - Trusted Health Information for You**
http://www.nlm.nih.gov/medlineplus/cysticfibrosis.html

**Cardinal Glennon Children's Hospital**
St. Louis, Missouri 314-577-5639
Website: http://pediatrics.slu.edu/index.phtml?page=geneticsdiv

**Children's Hospital at University Hospital and Clinics**
Columbia, Missouri 573-882-6991
Website: http://www.genetics.missouri.edu/

**Children's Mercy Hospital**
Kansas City, Missouri 816-234-3290
Website: http://www.childrens-mercy.org/content/view.aspx?id=155

**St. Louis Children's Hospital**
St. Louis, Missouri 314-454-6093
Website: http://www.peds.wustl.edu/genetics/